

This document is not part of the Invitae® clinical report and does not represent medical advice. These are general guidelines that are not specific to your result and may not represent all relevant international recommendations. You can use this guide to talk to your healthcare provider about your test results, clinical history, and the most current guidelines. This guide may not be appropriate for results that are suspected to be blood-limited, possibly mosaic, or suggestive of a larger imbalance of genetic material. We recognize that individuals have diverse gender and sexual identities. In this guide, the terms female, male, women, and men refer to sex assigned at birth.

What is a positive RPS20 result?



A positive test result means that a genetic change (variant) was found in the RPS20 gene. A positive RPS20 variant is considered “pathogenic” or “likely pathogenic” because it is associated with an increased chance for colorectal cancer.

What does this mean?



Individuals with a positive RPS20 variant have a higher chance of developing colorectal cancer than the average person. There may also be an increased chance for a bone marrow failure condition called Diamond-Blackfan anemia, however, lifetime risks are not clear. RPS20-related colorectal cancer can appear with few, or no precursor polyps. Colorectal polyps are noncancerous growths but some types can develop into cancer if they are not removed. While the chance for colorectal cancer is increased, the lifetime risk is not clear. Age of onset can vary and some individuals may never develop cancer. See the table later in this guide for more information and possible next steps.

What does this mean for family members?



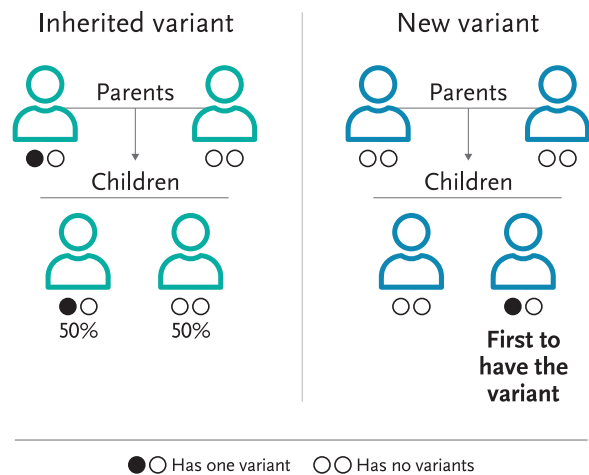
Relatives should be informed about these results. It is recommended that family members talk with their own healthcare provider about a plan for genetic testing and/or health screening. Genetic testing is a personal choice and some individuals may choose not to have genetic testing. Laws protecting employment and health insurance may apply to individuals undergoing genetic testing (for example, the Genetic Information Nondiscrimination Act in the United States).

Will family members have the same variant(s)?

The image shows where a RPS20 variant may have come from. Any individual can inherit and pass on a RPS20 variant, regardless of sex.

RPS20 variants can be inherited from a parent or an individual may be the first person in the family to have a new RPS20 variant. Genetic testing of parents may help determine the risk to the individual's siblings and other relatives. Individuals with a RPS20 variant can pass it on to children.

For individuals who are planning a family, reproductive options may be available to help lower the chance of passing on a variant to children.



Create a plan with a healthcare provider



These options are a guide for an individual and their healthcare provider. They are meant to be used along with an individual's genetic test results and other health information as part of a discussion to make a personalized care plan. Available options should be considered on an individual basis. A positive test result on its own cannot predict how a condition may affect an individual. This guide may not be appropriate for results that are suspected to be blood-limited, possibly mosaic, or suggestive of a larger imbalance of genetic material.

Options to consider

TOPIC	OPTION	MORE INFORMATION
Colorectal cancer	<ul style="list-style-type: none">Colonoscopy every 5 years beginning at age 20 (1)	<ul style="list-style-type: none">For individuals with a hematopoietic cell transplant prior to age 20, colonoscopy is recommended to begin one year after transplant (1)

These options include recommendations from NCCN (1). Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, and Gastric Version 1.2025 (1). © National Comprehensive Cancer Network, Inc. 2025. All rights reserved. Accessed 06/24/2025. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way. More information about genetics and disease continues to be available, so please always refer to the current guidelines and recommendations when considering surveillance and treatment options. Information on this document may not include all relevant international recommendations and acts as a supplement to the Invitae result report. This information is not meant to replace a discussion with an individual's healthcare provider and should not be considered or interpreted as medical advice. Additional resources provided within this document do not indicate or imply any endorsement by Labcorp with respect to any third party or any website or the products or services offered by any third party.

Resources



Genetic counseling can help individuals understand their genetic test results and options for next steps. Reviewing test results with a genetic counselor or other healthcare provider is recommended. Local or telehealth genetic counselors can be identified using the Find a Genetic Counselor search tool at nsgc.org (US and Canada).

Individuals with an Invitae test result can also log in to their patient portal (invitae.com) to view their results, contact a genetic counselor, or join the Invitae Patient Insights Network (PIN) (pin.invitae.com), an online platform where individuals can share information about their health and experiences to help advance research and drug development.

Notes for personalized assessment